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Case Report

LIFE WITHOUT LIMBS: TETRA-AMELIA

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ABSTRACT

Amelia is defined as the complete absence of the skeletal parts of a limb. Tetra- amelia, is the absence of all the four limbs is a very rare condition with an incidence range of 1.5–4/100,000 births. Many factors causing various limb defects are genetic, environmental (teratogens), vascular compromise by amniotic bands and conditions like oligohydraminos and maternal diabetes. Point mutation in the WNT gene plays a major role in causing limb defects.

KEY WORDS: Amelia, Tetra- amelia, Genetic, Teratogens, Maternal diabetes, Oligohydraminos.

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INTRODUCTION

The most critical period of limb development is from 24 to 36 days after fertilization [1].Limb defects occur in approximately 6 per 10,000 live births. Exposure to teratogens before day 33 results in serious limb defects .It may include major defects like amelia(complete absence of limbs), meromelia (partial absence), phocomelia (rudimentary hands or seal limbs) and minor defects like macrodactyly (overgrowth), polydactyly (duplication) and micromelia (all the segments of the extremities are shorter).Factors causing various limb defects are genetic, environmental (teratogens), vascular compromise by amniotic bands and conditions like oligohydraminos and maternal diabetes.

CASE REPORT

A full term, dead female foetus was obtained at the department of anatomy, MKCG medical college, Berhampur from CHC Badagada of Ganjam district in Odisha. On examination the baby had absence of all the four limbs. She was born to a couple of non- consanguineous marriage. Mother was non-alcoholic and non-diabetic with no history of radiation exposure.

Findings: On external examination following was observed as shown in figure 1.

Head & Neck

1.Cranium normal.

2.Face –Unilateral right upper cleft lip and cleft palate

3.Tongue ,Ear,Nose,Eyes -normally developed

Limbs:

4. Skeletal parts of the upper and lower limbs-Completely absent

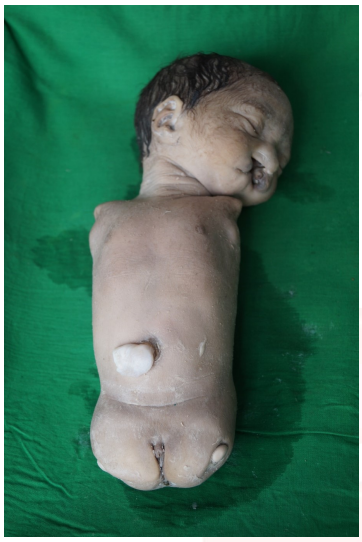
G.I.System

5.Intact anterior and posterior abdominal wall.

6.Rudimentory external genitalia, atresia of vagina.

7. Anus normally developed with presence of meconium.

Fig. 1: Baby with absence of upper and lower limbs(Tetra-amelia) associated with cleft lip and cleft palate.



DISCUSSION

Cases of Amelia has been reported in the literature but there are very few cases of tertra-amelia recorded till date. We are presenting a case report of an dead infant with tetra-amelia i.e absence of all the four limbs. Smithells (1962) has reported several cases of tetra-amelia. Amelia with Robert syndrome has also been reported by Song et al [2]. Amelia is found to be transmitted in some Arabs and Turkish families in an autosomal recessive manner. The etiology of Amelia is thought to be sporadic, yet its genetic basis cannot be ruled out. Different modes of inheritance like autosomal recessive, X-linked dominant and autosomal dominant indicates the genetic heterogenicity of the condition [3].

Research in animals show that WNT gene is critical for limb outgrowth. Wingless type MMTV, which is a part of a large family of WNT genes plays a major role in development before birth. It determines the antero-posterior axis during the earliest embryonic stage. It also helps in normal formation of facial features, head, heart, lungs, neurons, skeletal and genitalia. Tetra-amelia syndrome is a health condition related to mutation in WNT3 gene. The gene is located in the 17q21.The mutation that occur in the WNT3 gene is the point mutation in a sequence of DNA that results in premature stop codon in the transcribed RNA. This mutation is referred as Gln83Ter and results in the production of abnormally non-functional version of the WNT3 protein [4]. Thus loss of the WNT3 protein disrupts normal limb formation before birth and leads to other serious birth defects associated with tetra-amelia syndrome.

In many cases, the lungs are under-developed which makes breathing difficult so that the babies are mostly stillborn or die shortly after birth [5].In our case also the twenty years old primigravida vaginally delivered the 2.1kg baby, who cried soon after birth but died twenty minutes later. Recurrence of Amelia is also reported in some families but in our case family history was non-contributory. Even cases of tetra-amelia are reported with a normal chromosomal finding.

CONCLUSION

This case is reported because of its rare occurrence so that proper awareness can be spread. Due to absence of antenatal check-up diagnosis was made only after the baby was delivered. The parents were counselled for a low recurrence rate and were advised for antenatal check-up as well as early anomaly scan in future pregnancies. Antenatal screening, karyotyping analysis, and genetic counselling are recommended to reduce the risk of such congenital anomalies.

Conflicts of Interests: **None**

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